



## Model of Care for Transition from Paediatric to Adult Healthcare Providers in Rare Diseases

The National Clinical Programme for Rare Diseases



National Clinical  
& Integrated Care Programmes  
*Person-centred, co-ordinated care*



**ROYAL  
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For further information about the National Clinical Programme for Rare Diseases please visit

[www.hse.ie/eng/about/Who/cspd/ncps/rare-diseases](http://www.hse.ie/eng/about/Who/cspd/ncps/rare-diseases)

## 1. EXECUTIVE SUMMARY

For rare diseases in Ireland, collaboration across various healthcare settings is key given the nature of national services and expertise. This requires system-level solutions which address the alignment of providers in multiple settings, with facilitated communication, record sharing, capacity building and related audit and clinical research nationally.

A rare disease is defined in the EU as a disease or disorder affecting fewer than 5 in 10,000 of the European population. There are an estimated 6-8,000 known rare diseases affecting up to 8% of the total EU population, representing up to 300,000 Irish people during their lives. Estimates suggest that around 50-75% of all rare diseases affect children. Recognising the vulnerable situation in which adolescents with rare diseases are often placed at the point in their healthcare where they need to transfer from paediatric to adult healthcare providers, a working group was established from the National Clinical Programme for Rare Diseases to develop a model of care for such individuals.

In this process it was established that transition in any condition, either a common chronic illness or an extremely rare condition, must be carried out in a planned, phased approach, which leads ultimately to seamless transfer of care as is appropriate to the patient's healthcare needs and developmental stage. In the future it is hoped that by developing centres of expertise where there can be concentration of expertise and resources, that patients with specific rare diseases can be managed in a more holistic pathway avoiding potential gaps in care or expertise, loss of access to allied healthcare services previously available to the individual, and loss to follow up during the transition phases.

Many challenges in transition are not unique to rare diseases, but certain aspects of a rare disease can bring extra hurdles which can be offset to a degree by involving the patient in the decision on where to refer on to adult care. For this reason it is particularly important that the patient with a rare disease is part of their care pathway from early adolescence such that self-advocacy, independent health care behaviour, and sexual health are addressed. Furthermore, patients with rare diseases should be offered psychosocial support which may include access to genetic counselling, advice on education and vocational planning, and general promotion of a healthy lifestyle. The transfer of information to the adult healthcare provider in a timely fashion is vital in the transition process. The transition pathway in rare diseases needs to recognise and incorporate the intellectual or physical issues facing the patient, and ensure that carers involved are also part of the process. In conditions where multiple systems are involved, it should be agreed in advance which physician involved in the care will be taking a lead in the management of the patient, and this physician should be appropriately resourced, where possible to allow a multi-disciplinary team approach to care.

Formal structured training for physicians in training and other healthcare providers on aspects of adolescent medicine, transition and rare disease would benefit patients with rare diseases during the transition process. Furthermore, as many patients with rare diseases can have problems with mental health which as a feature of the condition can deteriorate in adolescence/ early adulthood so that extra resources and access to mental health/ clinical psychology services should be available.

Also, many patients with rare diseases have life-limiting conditions and the potential need for palliative care and respite care for carers should be planned for and addressed during the transition process so that abrupt breaks in these aspects of the care are avoided.

In summary, it is hoped that this document can achieve the goals of transition to ensure that young people affected with rare diseases approaching transition 'get the right care, at the right time, and in the right place' via enhanced communication between paediatric and adult healthcare providers well in advance of transfer of care, engaging the patient/ carer in the decision, and facilitating access to expert multi-disciplinary care in a particular rare disease where available, regardless of location and resourcing centres of expertise for management of rare diseases nationally.

## 2. INTRODUCTION TO TRANSITION IN RARE DISEASES

### 2.1 DEFINITIONS

A rare disease is defined in the EU as a disease or disorder affecting fewer than 5 in 10,000 of the European population. There are an estimated 6-8,000 known rare diseases affecting up to 8% of the total EU population, representing up to 300,000 Irish people during their lives. Estimates suggest that around 50-75% of all rare diseases affect children <sup>(1)</sup>.

An ultra-rare disease is generally considered as that with a prevalence of less than 1 per 50,000 population, therefore in the Irish context diseases for which there are less than 100 confirmed cases nationally would be considered ultra-rare <sup>(1)</sup>. With ultra-rare conditions awareness among medical professionals may be limited as most physicians regardless of specialty may only see one or two such patients during their career. Resulting unfamiliarity with the condition may lead to numerous misdiagnoses, delay in diagnosis, and sub-optimal treatment.

### 2.2 TRANSITION

The concept of transition was defined by Blum et al in 1993 as the “purposeful and planned movement of adolescents and young adults with chronic physical and medical conditions from child-centred to adult oriented health care systems” <sup>(2)</sup>.

The American Academy of Pediatrics (AAP), American Academy of Family Physicians (AAFP) and the American College of Physicians – American Society of Internal Medicine (ACP-ASIM) 2002 Consensus Statement describes transition as the “process to maximise lifelong functioning and potential through the provision of high-quality, developmentally appropriate healthcare services that continue uninterrupted as the individual moves from adolescence to adulthood” and the AAP has developed a number of transition resources including ‘Got Transition’ (<http://www.gottransition.org/>)<sup>(3)</sup>.

Young individuals affected with rare diseases, with the additional burden of having complex, unrecognised conditions, are in great need for a nationally agreed model of care designed to address safe progress of adolescents through our medical system. Current structures of delivering health care for large numbers of patients with common conditions do not usually suit patients with complex rare diseases, especially those with multisystem involvement. Therefore the period of transition of care is a particularly vulnerable stage in medical management.

A central recommendation of the National Plan for Rare Diseases (2014-2018) is the development of the clinical and organisational governance framework that will underpin care pathways and access to treatment for rare disease patients, particularly in the context of the transition from paediatric to adult care.

Although improved therapies and recognition of rare diseases has led to improvement in both quality of life and longevity for some people with rare diseases, this means that more children with rare diseases are ‘graduating’ to adult services. There is a great need for coordinated care for young people with rare diseases as these conditions are frequently multisystemic and complex with often fragmented care pathways. Furthermore, there may be limited specialty experience available for

many of these conditions in adult practice. Research within the disability sector also shows that reaching the age of 18 is a precarious time in terms of finding appropriate adult services, e.g. medical, educational or social, and in terms of finding appropriate employment.

### 2.3 SCOPE OF THE MODEL OF CARE

A sub-group of the working group of the National Clinical Programme for Rare Diseases has developed this Transition Model of Care and Recommendations to facilitate and progress the Transition Process for rare disease patients moving from a paediatric to an adult care hospital setting. Due to the large number of rare diseases, this document is designed to be generic rather than specific to one condition or group of conditions. The goal is to provide a framework from which dialogue can begin and to raise awareness of the issue of transition of patients with rare diseases and their broad range of needs as a whole, and to distinguish these patients' needs from those of patient with more common chronic conditions who benefit from 'strength in numbers'. Although reference is made to educational and vocational aspects of life for a person with a rare disease, this is intended as a clinical document. We acknowledge the huge role played by healthcare professionals, allied healthcare workers and other state agencies in the community, along with patient representative groups, however specific reference to these roles is beyond the scope of this document.

### 2.4 EUROPEAN AND POLICY BACKGROUND

In response to the European Commission 2009 recommendations on action in the field of rare diseases, Ireland published its first national plan for rare diseases in July 2014. The need to provide a transition care pathway for adolescents with rare diseases to the appropriate adult services has been recognised as a major recommendation in the 2014-2018 national plan<sup>(1)</sup> following the 2012 Rare Diseases Public Consultation and the summary recommendations of the 2011 Europlan consultations (incorporating the views of 15 Member States) wherein the issues of transitioning from paediatric to adult services was identified as a major challenge. It was proposed that Case Managers could be identified to dedicate their attention to individual cases and follow them all along the healthcare pathway including the transition from the paediatric to adult age, thus providing integrated care.

In 2016 NICE published guidelines<sup>(4)</sup> on transition, stating that "Transitional care should become a shared priority despite the pressures on current funds".

Integrated care is defined by the World Health Organisation as "a concept bringing together inputs, delivery, management and organisation of services related to diagnosis, treatment, and care, rehabilitation and health promotion". Irish parents of children with rare diseases patients seek access to healthcare that is seamless, smooth and easy to navigate and a coordinated service that enables the patient to move from the paediatric to adult setting.

## 2.5 RARE DISEASE CENTRES OF EXPERTISE

The emerging role of Rare Diseases Centre of Expertise both in Ireland and across Europe will have a key role in facilitating integrated care provision in line with the EUCERD recommendations on quality criteria for Centres of Expertise in Rare Diseases<sup>(5)</sup>. Centres of Expertise bring together or coordinate, multidisciplinary competences/skills, including paramedical skills and social services within the specialised healthcare sector.

Centres of Expertise should play a key role in facilitating integrated health care provision by bringing together or coordinating multi-disciplinary teams. The European Commission 2009 recommendations in the field of rare diseases are that Member States should promote multi-disciplinary team working, holistic approaches, continuous person centred and participatory care in both health and social care.

Care for rare diseases patients in essence then should be integrated at all levels and in particular with primary care services so that patients and families can receive care as close to home as possible. International research into primary care services firmly relates to the unique role of the General Practitioner (GP) in terms of continuity and coordination of care and the resources provided to support GP care<sup>(6,7)</sup>.

## 2.6 MEDICAL ADVANCES AND THE IMPACT ON RARE DISEASES

Advancing medical treatments and understanding of the pathophysiology have dramatically changed the management, quality of life and life-expectancy of many people with rare diseases. The establishment of registries and national and international networks for sharing of knowledge and experience has impacted on many conditions where a cure or survival beyond early childhood was previously not possible. Earlier diagnosis due to national or targeted screening programmes, advances in genetic testing and more understanding of the variations in phenotype and the spectrum of disease ensures that treatment can be tailored to individuals to prevent complications, improve quality of life and delay the natural history of the disease (e.g. cystic fibrosis, neonatal diabetes, congenital hyperinsulinism, mucopolysaccharidosis, cystinosis).

Conditions previously considered universally fatal have now reached over 90% survival in the past 50 years (e.g. severe combined immunodeficiency) and many children now survive into adult life with complex chronic rare diseases that previously did not survive past infancy (e.g. severe osteogenesis imperfecta, Inherited Metabolic Diseases such as Lysosomal Storage Disorders, Glycogen Storage Disease, mitochondrial disorders, amino acidopathies, organic acidaemias, and urea cycle defects) with coexistent increasing medical complications requiring management. Conditions such as Duchene Muscular Dystrophy (DMD) until two decades ago associated with inevitable demise in the second decade now survive into the third and fourth decade<sup>(8,9)</sup>. In other conditions, advances in assisted human reproduction have seen the development of new complications (e.g. Turner syndrome and aortic dissection in pregnancy).

Overall, the population of adults with rare conditions will likely continue to grow. Increased survival of children with such conditions means that many childhood conditions have now also become adult diseases. Currently, the vast majority of children with congenital or chronic rare diseases survive to adult life so that although it is often cited that over 70% of rare diseases present in childhood, there



is an exponential increase of the prevalence of individuals affected with rare diseases over time and into adulthood. Paediatric conditions are no longer confined to childhood but the disease-specific expertise may currently be predominantly amongst paediatricians familiar with their management. There is an increasing need for medical transfers from paediatric to adult services including a need for genetic counselling for specialist clinics treating rare diseases, and where appropriate, a focus on genetic counselling for the patient who is transitioning to adult services <sup>(10,11)</sup>.

Furthermore, there is increasing evidence to suggest that in rare diseases where there are small numbers affected in a population (in particular ultra-rare conditions) that condition specific high quality, patient centred clinics are the best model for co-ordination and provision of care which may require extra resources but should ultimately be more cost effective <sup>(12)</sup>. Some models are currently set up in other countries (e.g. Birmingham UK - Alstrom syndrome <sup>(12)</sup>, Germany – Bavarian cystinosis programme, France - Silver Russell and Prader Willi syndromes).

The increased survival of complex, life-limiting rare conditions brings challenges for physicians, health and social care practitioners and carers. These range from the impact on educational and social resources for minor impairment to learning, to those with sensory impairments and to those associated with more long-term survival and the need for palliative care.

### 3. PLANNING FOR TRANSITION IN RARE DISEASES

#### 3.1 INTRODUCTION

For successful transition to occur a developmentally appropriate transition plan should be developed in collaboration with young person and their family. The transition plan should address not only the young person's specific health issues but their wider physical, developmental, psychosocial, mental health, educational, lifestyle, cultural and financial needs <sup>(14)</sup>. Six key areas have been identified as the core issues for discussion throughout the transition process to ensure a comprehensive and holistic encounter for the participating adolescent and their families <sup>(15)</sup>. These are:

1. self-advocacy
2. independent health care behaviour
3. sexual health
4. psychosocial support
5. education and vocational planning
6. health and lifestyle

Consequently, transition should consist of joint planning with youth and parents/caregivers to foster independence and active participation in decision-making. It also consists of assistance in identifying adult providers and ensuring a smooth transfer to adult-centred care with current medical nursing and therapeutic information. In this context, it is considered that the transition from a paediatric to an adult oriented service should not be a sudden unanticipated transfer, but should be an organised integrated process of preparation and adaptation. To this effect, transition has often been cited as the most important event in the care of patients with chronic diseases, and effective transition has been shown to improve long-term outcomes and the experience of the young person <sup>(16-19)</sup>.

#### 3.2 CURRENT CHALLENGES IN TRANSITION

The various unique challenges in transition include the fact that there is no "one-size fits all" programme, and there is currently no formal training in Adolescent Health for Irish trainees through the Royal College of Physicians of Ireland (RCPI) Higher Specialist Training programme in General Paediatrics <sup>(20)</sup>, which is in contrast to programmes in the UK and Australia <sup>(21, 22)</sup>. In addition there are various misconceptions of the transition process which can be confused with transfer of care <sup>(23)</sup>, such that infra-structural planning and co-ordination between services at different locations can be challenging (e.g. the two main paediatric hospitals in Ireland are not co-located with adult services). Furthermore all health professionals working within a multi-disciplinary team treating patients with rare diseases could benefit from training or education in the concepts of both Adolescent Medicine and the challenges facing patients with rare diseases to enhance focus in this area <sup>(24)</sup>.

### 3.3 ADOLESCENT DEVELOPMENT AND ADHERENCE

Adolescence describes various age ranges from as young as 10 and up to 24 years but is defined by The World Health Organization as a period between 10-19 years<sup>(25)</sup>. After infancy, adolescence is the period of greatest and most rapid development, however compared to other phases of life such as infancy, it is relatively understudied in relation to future health<sup>(26)</sup>.

Young people face many challenges when preparing for and entering adult life regardless of their health. The transition period from childhood to adulthood is already a time of great physiological, psychological and social change coincident with changes in schools, education, work and employment, new challenges and specific social hazards and pressures such as exposure to illicit drugs, alcohol misuse and other risk taking behaviour. When also faced with complex physical health needs, there are many additional hurdles<sup>(27)</sup>. Often the health needs of these young people will have been met by the same people who have looked after them for as long as they can remember where a strong relationship of trust and dependency has been formed over many years. One of the biggest changes such young people face as they anticipate adulthood is the transfer to a health care environment where they may now need to consult with several different health care teams and services, and the reality of starting on a whole new journey and establishing new relationships.

In addition, adherence issues can be very problematic in adolescent and young adult life. 'Adherence' refers to the extent to which patients follow medical treatment and advice. The combination of rare genetic conditions and adolescence adds a whole new range of non-adherence factors, e.g. having to access specialist care centres, and treatment regimens involving different systems, lack of insight into their condition, and lack of availability of a support person who was always present through childhood (i.e. the parent/guardian).

The developmental cognitive and behavioural challenges in adolescence have an influence on transition<sup>(28-29)</sup>, with the period of change in the adolescent brain extending from 11 to 25 years of age<sup>(30-31)</sup> and the need to change from paediatric health care to the better match in the adult health care environment<sup>(33)</sup>. There is a broad age range at which this may occur however, depending on the condition, the cognitive abilities of the patient and the support structures from and to which they are transitioned.

### 3.4 ALIGNMENT WITH PUBLISHED MODELS OF CARE FOR IRISH HEALTH SERVICES

The National Clinical Programme for Paediatrics and Neonatology recommends that each service should have its own policy on the timing of transfer<sup>(34)</sup>. Although there is no existing general transition policy in place in Ireland, the National Clinical Programme for Paediatrics and Neonatology plans to develop a national guideline for transition in chronic disease. The general suggestion is that between the ages of 16-18 years transfer of care from paediatric to adult services should occur. 'Rigid age limits defining children's and adult services are not desirable as the timing must depend on the developmental readiness and health status of the individual adolescent as well as the capabilities of the adult providers'. The time since completion of treatment might be relevant in some cases, and transition, where possible, should be during remission rather than relapse or active disease. Furthermore the National Clinical Programme for Neurology has recently highlighted that transition should be a planned phased process and include a suggested checklist<sup>(35)</sup>. However, chronic illness

and its treatment can result in both physical and psychological delay in maturation, therefore the developmental stage of the individual needs to be considered in this process <sup>(36)</sup>.

The Model of Care for Epilepsy published by the National Clinical Programme for Epilepsy also outlines the necessity of transition “to achieve the best outcomes possible for patients and their families in areas of health, independence and adulthood” <sup>(37)</sup>. The model of care also notes the benefits of transition clinics which introduce patients with epilepsy to the staff on the adult service.

### 3.5 TRANSITION IN CHRONIC DISEASE

Despite our increasing knowledge of the importance of transition there are many barriers to successful development and implementation of transition policies. These barriers range from structural to cultural to logistical. Along with a variation in the location of services from standalone paediatric units to tri-located units, a lack of suitable information technology supports make consistent implementation challenging. These challenges and deficits in turn result in a lack of confidence in adults services for the young person, their families and the providers. This may lead to disengagement of the young person, loss to follow-up and increased morbidity and mortality around the transition years and into early adulthood. A recent quantitative study of perceptions of transitional care highlights that adolescents, parents and providers are dissatisfied with aspects of the care delivery process in transitional care itself with only small differences in disease-specific issues. Specifically when asked about opportunities to make their own decisions, adolescents rated current care significantly worse than their parents did. Similarly, when adolescents were asked about the social skills of their treating clinicians or about being seen by clinicians without their parents present their ratings were significantly worse than those of their parents <sup>(38)</sup>.

The transition from paediatric to adult care, especially when poorly planned, is noted to be associated with poor clinical outcomes, increased costs and low patient and family satisfaction as well as increased healthcare costs associated with emergency department visits, hospitalisations, and intensive care admissions <sup>(39), (40)</sup>. Changes in circumstances, (university, work, financial pressures etc.) may mean that the original intended adult service may no longer be convenient to the patient, and alternative permanent or temporary adult services may be required. Patients and providers thus need to be aware of the differences between adult and paediatric care and institutional support must be provided to ensure follow up.

Poorly planned transition from paediatric to adult health services along with psychosocial factors is associated with an increased risk of non-adherence to treatment and loss to follow up in chronic conditions <sup>(41, 42)</sup> resulting in serious consequences - both in terms of morbidity <sup>(43-48)</sup> and mortality <sup>(49-52)</sup>. Challenges identified include the shift in input from parents, differences in focus during the patient consultation between paediatric and adult clinics, disengagement by the adolescent patient and risk taking behaviours which increase the risk for immediate and long-term complications <sup>(48)</sup>. There is increasing evidence that following the move from paediatric to adult services, morbidity and mortality increase for young persons with chronic diseases such as Type 1 Diabetes Mellitus (T1DM) <sup>(47,49)</sup>, post renal transplant <sup>(43)</sup>, congenital heart disease <sup>(53)</sup>, and neurological conditions <sup>(54)</sup> which can also be associated with poor social outcomes and educational achievement.

When a chronic disease is stabilised in the paediatric phase, transition and early adulthood care brings forward the importance and central position of long-term psychosocial aspects, often more challenging than the primary medical issues. There is frequently a change of focus from staying alive in the paediatric years to now dealing in adolescence and young adult life with the burden of chronic conditions, with evolving or risk of multisystem complications and sometimes palliative care issues if the disease is life-limiting. At the same time, the young adult is also faced with the challenges of attempting to succeed in independent living, of dealing with issues relating to housing, life insurance, disability allowances, obtaining further education, employment, ageing carers, contraception, and frequently complicated pregnancy and fertility issues.

Successful transition planning therefore depends on careful collaboration between child and adult services. The timing of transition is paramount, however from diagnosis there should be a plan for the future of the child that begins with the end of paediatric care in mind. Transition planning should include linking families and young adults in order to share information and experiences and encouragement of the child/young adult to accept responsibility for his or her own health care <sup>(42)</sup>.

### 3.6 CHALLENGES FOR TRANSITION IN RARE AND ULTRA-RARE DISEASES

In the context of rare and ultra-rare diseases this process has added and unique challenges but, three groups with broadly different needs should be considered in the planning process:

1. Those with a rare chronic but relatively stable condition for which normal or near-normal life expectancy is anticipated.
2. Those with a rare life-limiting condition.
3. Those with a rare condition associated with complex needs - intellectual and/or physical disability.

Especially in the latter two groups, the additional needs of caregivers around the time of transition need to be considered.

Currently, good examples of well-developed transition care pathways from paediatric to adult care exist, illustrated by a number of rare diseases available on Orphanet <sup>(55)</sup> e.g. the EURO-WABB site <sup>(56)</sup> with a guideline available for the rare diabetes syndromes, Bardet-Biedl, Alstrom and Wolfram Syndrome and also the “Treat and Care” NMD sites for Duchenne Muscular Dystrophy <sup>(57)</sup>.

By virtue of their rarity, patients with rare diseases are frequently small in number and therefore lack the “strength in numbers” or “critical mass” associated with other more common conditions where dedicated multi-disciplinary clinics can be supported. Furthermore the empowerment and voice of patient advocacy organisations may not exist for many conditions, and those in existence often compete for resources and access to policy makers with those representing more common conditions.

As rare diseases are often associated with greater complexity, a wide range of medical problems and the need for access to more than one physician or specialist, even at multiple different sites in different healthcare settings can ensue, especially as the patient gets older and an increasing number of complications of the conditions arise such as neurocognitive decline, psychiatric disorders <sup>(58,59)</sup>, respiratory compromise, reduced mobility, osteoporosis, metabolic and sleep problems <sup>(60,61)</sup>. This

multiplicity of health care needs is particularly challenging in rare diseases where a defined care pathway may be lacking, and continuity of care is key<sup>(62)</sup>. In many conditions the imminent or eventual need for palliative or end of life care may need to be addressed (see Flow Sheet 2).

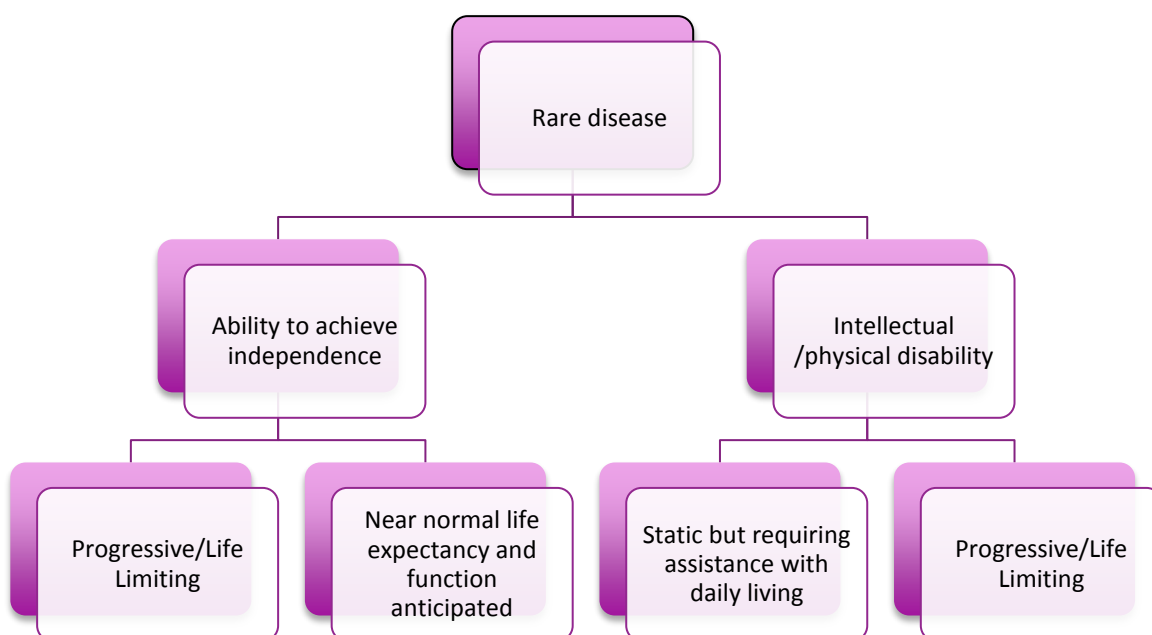
## 4. ADDITIONAL FACTORS TO CONSIDER IN RARE DISEASES TRANSITION

### 4.1 DISABILITY, PSYCHOSOCIAL NEEDS AND MENTAL HEALTH

Many rare diseases are associated with significant intellectual or physical disability, which may complicate the transition process (see Figure 1). Furthermore, the increasing psychosocial needs and challenges of adolescence are known to impact on the transition process <sup>(42)</sup>. It is well recognised that children with special health care needs are less likely than their peers without disabilities to succeed at some transitions. They are less likely to graduate from secondary school, attend college, work in competitive employment or live independently. Many have little or no experience managing their own health care, making medical appointments or discussing their medical conditions. Also their carers often have low expectations about their prospects of future employment and independence <sup>(63,64)</sup>.

Reduced quality of life in chronic illness is a recognised problem in both childhood, adolescence and adulthood <sup>(65)</sup>. The impact of a rare disease on patients and families ranges from minor inconvenience to extreme disruption to family life. The impact of certain conditions on quality of life has been widely studied in a variety of rare conditions with varying results depending on the type, severity and impact of the condition on normal daily living <sup>(65-70)</sup>. A number of rare conditions are also associated with deterioration in cognitive function, or decline in social functioning as adolescence progresses e.g. Prader Willi, Klinefelter, Williams and 22q11.2 deletion syndromes <sup>(59,60)</sup>. Therefore anticipation and preparation for declining mental health in early adulthood should begin during childhood before this impacts on the transition process and adherence to treatment.

FIGURE 1. DISTINCTIONS IN RARE DISEASE – BASED ON NEUROCOGNITIVE IMPAIRMENT/PHYSICAL DISABILITY, CARE NEEDS AND LIFE EXPECTANCY



## 4.2 LIFE-LIMITING CONDITIONS

A life-limiting condition is defined as any illness in a child in which there is no reasonable hope of cure and from which the child or young/middle aged adult will die. Whilst the majority of children with life-limiting conditions are unlikely to live beyond 18 years, some whose diagnosis is made in childhood will survive unexpectedly into early adulthood. While many rare conditions have no impact on long term prognosis, some are life limiting from diagnosis and some rare diseases can become life-limiting over time.

The transition process for these young adults can be a stressful time due to a lack of emotional support<sup>(71)</sup>, paucity of information<sup>(72)</sup>, challenging issues related to inpatient experiences<sup>(73)</sup>, and a loss of services such as respite breaks when in adult care<sup>(74)</sup>. There are few models of good practice in terms of transition services for adolescents with palliative care requirements<sup>(75)</sup>.

Studies specific to Ireland on this topic indicate that positive outcomes of the transition process for young adults with life-limiting conditions depend on adequate resourcing, equipping and preparation of adults services with skilled knowledgeable staff capable of providing continuity of care, allowing the young adult to become increasingly responsible for their medical management, and the young adult feeling “transition ready”<sup>(76-79)</sup>. Furthermore, there is an increasing sense of empowerment in adult service providers when information is received about the young adult and their ongoing involvement in the transition process.

Palliative care for children is an active and total approach to care, embracing physical, emotional, social and spiritual elements<sup>(80)</sup>. Many children requiring palliative care have life-limiting conditions, and it is recognised that children may survive many years with these conditions, and that the needs of these children differ from the needs of adults<sup>(81)</sup>. With increased survival of children with certain life-limiting conditions into adolescence, palliative care may become a reality during adolescence, temporally coinciding with the time of transfer of care from paediatric to adult services. The 2005 Paediatric Palliative Care Needs Assessment co-funded by the Department of Health and the Irish Hospice Foundation advocates a coordinated approach to age appropriate care and was followed by a national policy<sup>(82-83)</sup> in 2010.

The importance of early involvement of paediatric palliative care has recently been emphasised<sup>(84)</sup>. The differences between adult and paediatric palliative care are the number of children involved, the rarity of conditions, the time-scale and the frequency of familial conditions. This has implications for counselling and recurrence risk, the need for care to be family-centred care in the context of ongoing child development and also for the family burden of care to be acknowledged.

Furthermore, given the frequently unknown prognosis, there is an awareness of the need for palliative care to embrace ongoing education of the child. Some children whose life expectancy is short may need intense involvement over a limited period of time. Others will require intermittent involvement at different stages of their illness before needing active continuous care when they reach the terminal phase. Hence, identifying when a palliative approach is appropriate can be challenging.

In addition, the nature of some life-limiting rare conditions in children makes predicting prognosis challenging. Some life-limiting illnesses can have relapsing remitting phases where many episodes of critical illness and near death can revert back to periods of relative stability and chronic illness before



final demise. This can result in a huge psychological burden on the caregivers, patient and family, and may coincide with periods where transfer of care has been planned or anticipated.

There are four categories of life-limiting conditions <sup>(84)</sup>:

1. Those for which curative treatment has failed e.g. cancer, irreversible organ failure
2. Conditions associated with periods of normal childhood activities, which usually require long periods of intensive treatment, but which are often associated with premature death e.g. cystic fibrosis, muscular dystrophy, congenital heart disease
3. Progressive conditions without curative option where treatment may ameliorate the condition, which may extend over a number of years e.g. Batten's disease, Mucopolysaccharidosis.
4. Conditions with severe neurological disability that may deteriorate unpredictably but are not usually considered progressive e.g. severe brain or spinal injury, severe cerebral palsy.

## 5. KEY POINTS FOR SUCCESSFUL TRANSITION

### 5.1 INTRODUCTION

The recommendations outlined in this chapter aim to develop a rare diseases national transition model of care, the application of a set of service principles across identified clinical streams/providers to ensure that young people affected with rare diseases approaching transition 'get the right care, at the right time, and in the right place'. A successful transition plan is a multi-faceted process that engages multiple care providers and accommodates a patient's condition, age and developmental stage. Key points to successful transition<sup>(24)</sup> include early preparation of the young adult for independence and self-advocacy<sup>(15)</sup>, an education programme for patient and parent that addresses medical, psychosocial and educational/vocational aspects of care that is age and developmentally appropriate, identification of a key worker, the development of a written transition policy agreed by all members of the multidisciplinary team and targeted adult services.

If transfer is to a distant clinic, the adolescent should receive detailed information and visit the adult clinic well in advance, preferably with a trusted carer, to introduce them to the adult environment and staff. The co-ordination of this process can be undertaken by a specific member of the multidisciplinary team caring for the young person, such as clinical nurse specialists or therapists, who can address patient concerns prior to formal transfer<sup>(34)</sup>. Communication and handover of the appropriate documentation is key with the option to transfer the complete patient file with consent of the patient<sup>(85-87)</sup>.

The plan for each patient created with the young person and their family should be regularly reviewed and updated with liaison personnel in both paediatric and adult teams. The presence of an interested, capable and adequately staffed adult clinical service is required with good communication throughout the process.

Examining the strategies most commonly used in successful programmes confirms the importance of patient education and skills training and of specific young adult clinics, either jointly staffed by paediatric and adult physicians or dedicated young adult clinics within adult services<sup>(39)</sup>. Care of the child with a chronic condition is largely family centred with the parents providing a very active role. At the time of transition, care in the paediatric setting must adapt to accommodate the emerging adult who needs to be treated independently of the family structure.

Transition should be a process of preparing the young person for the adult service, which should start at a young age 11+<sup>(23)</sup> or at least by 14 years<sup>(3, 28, 88)</sup> rather than one single event. A key accountable individual responsible for supporting their move to adult health services has been strongly advocated as a means to support a smooth transition process.

The success of the transition process will also depend on formal systems that ensure accessibility and transfer of information, including a portable and accessible up-to-date medical summary such as a 'health passport' to ensure relevant professionals have access to essential information about the young person. Children's services should be the providers until adult services take over. It is paramount and should be confirmed with the adult provider that the paediatrician will be responsible for continuity of care until the young adult is seen and transferred to the adult setting.

**TABLE 1: RECOMMENDATIONS FOR SUCCESSFUL TRANSITION OF A YOUNG PERSON**

The recommendations in this table include the six core elements of Health Care Transition supported by The American Academy of Paediatrics/American Academy of Family Physicians/American College of Physicians which include a number of tools for assessment <sup>(89)</sup>.

<p><b>General transition Recommendations</b></p>	<ol style="list-style-type: none"> <li>1. Transition should be a process rather than one single event. The young person should have attended at least one (but ideally several) clinic with the adult team in attendance prior to transfer</li> <li>2. Children’s services are the providers until adult services take over</li> <li>3. Avoid if at all possible transfer of care during a period of acute illness or when a patient is dying</li> <li>4. Transition and young adult clinics should ideally be held at a convenient/suitable time for the young person that does not clash with other commitments. Long waiting times should be avoided to encourage attendance</li> <li>5. The care pathway and medical summary should be available for each patient or as an appropriate transfer of the patient file with obtained consent</li> <li>6. A transition plan, written by a healthcare professional that is familiar with the patient's care to date is written (ideally by age 14).</li> <li>7. Encourage patient to participate in evaluating their self-management</li> <li>8. Hospital transition policy with input from youths and families that describe the practice’s approach to transition, and educate the staff about transition</li> <li>9. Conduct regular transition readiness assessments, beginning at age 11-14 – plan transition, develop and update care plan</li> <li>10. A key accountable individual responsible for supporting the move to adult health services</li> <li>11. Formal systems for accessibility and transfer of information, e.g. ‘health passport’</li> <li>12. Health services provided in an appropriate environment that takes account of their needs, without gaps in provision between child and adult services</li> <li>13. Youth focused and family-centred, developmentally and culturally appropriate expert care in adult facilities</li> <li>14. Training and advice to prepare for transition to adult care including consent / advocacy</li> <li>15. Adequate access to independent advocates for young people</li> </ol>
<p><b>Ideal for rare diseases</b></p>	<ol style="list-style-type: none"> <li>1. Establish criteria for identifying transitioning youth and enter their data into a registry</li> <li>2. Care is provided by staff with expertise in the particular condition</li> <li>3. A transition policy is developed for the service</li> <li>4. A service-specific transition co-ordinator support patients to transition from paediatric to adult healthcare services</li> <li>5. Respite/short break facilities are available</li> <li>6. Carers’ needs are assessed</li> </ol>

## 5.2 EXISTING TRANSITION PROGRAMMES IN IRELAND

In the Irish setting, the transition issues affecting young adults and adolescents with Type 1 Diabetes Mellitus have been well described by the National Clinical Programmes for Diabetes and Paediatrics although the model recommended has yet to be broadly implemented.

For rare diseases, examples of a number of very successful transition programmes are clearly accessible on the “stepping up” website <sup>(90)</sup> which are outlined in Table 2.

The YARD (Young Adolescents with Rheumatic Diseases) programme run between the National Centre for Paediatric and Adolescent Rheumatology at OLCHC and the Rheumatology Department at St Vincent’s University Hospital provides a unique and seamless transitional pathway of care for adolescents and young people with rheumatic conditions (including rare rheumatological conditions). This bimonthly combined adult and Paediatric clinic commenced in 2006 and is a dedicated service for those patients graduating from mid to late transition from Our Lady’s Children’s Hospital, Crumlin as well as teenagers/young adults presenting to medical services for the first occasion in the latter teen years. The clinic is supported by an Adolescent CNS and an Adult Rheumatology ANP as well as other MDT members and AHPs. The planned transitional care is age-appropriate and is an active ongoing gradual process. The programme also provides support for parents and carers to assist in addition to encouraging by the young person to become more independent in their decisions and choices, and it also focuses on education for health care professionals. Ideally patients who complete the late transition (16-19 years) stage gradually transfer their care into a dedicated adult clinic that is provided by the same Adult physician and adult ANP for continuity of care. Furthermore the National Centre for Hereditary Coagulation Disorders has a long-standing seamless transition process. Importantly also, both of these services clearly identify adult clinicians to co-ordinate future care.

Congenital Heart Disease now represents the largest number of birth defects in Ireland, accounting for 1% of all live births per year. As 95% of children now survive into adulthood, The Mater Misericordiae University Hospital has developed an Adult Congenital Heart Disease (ACHD) service in the last five years. This national service provides specialist support to patients with complex congenital heart defects requiring lifelong care, with approximately 250 new patients transferring each year.

The successful Irish transition programmes are however currently mainly disease-specific and often institution specific and further development remains burdened with capacity and funding barriers. Regardless, their success relies on good working relationships and collaboration between the respective paediatric and adult sub-specialists.

TABLE 2: NATIONAL MODELS OF TRANSITION

Model	Service description	Examples in Ireland <sup>1</sup>
Dedicated follow-up service	<p>Provided within adult setting. No combined paediatric-adult clinic.</p> <p>No direct input or continuity from paediatric services</p>	<ul style="list-style-type: none"> <li>T1DM in most centres</li> </ul>
Seamless clinic	<p>A clinic, which begins in childhood/adolescence and continues into adulthood, with both paediatricians and adult physicians providing care in MDT as appropriate.</p> <p>Duration of combined care can vary from individual to individual</p>	<ul style="list-style-type: none"> <li>YARD – OLCHC and SVUH</li> <li>Cystic fibrosis – SVUH/OLCHC AMNCH</li> <li>Combined Rare Cardiac Genetic conditions (Adolescent and Adult) AMNCH</li> <li>Hereditary Ataxia Clinic (Adolescent and Adult)</li> <li>Cardiac arrhythmias and rare cardiac congenital disorders- MMUH</li> <li>Coagulation disorders/ haemoglobinopathy OLCHC and St James Hospital</li> </ul>
Life-long follow-up within paediatric setting	<p>Common default option especially in the past – often used in patients with life-limiting conditions, complex patients e.g. spina bifida due to parents/physicians who may be reluctant for transfer of care</p>	<ul style="list-style-type: none"> <li>Spina bifida</li> <li>Life-limiting disorders</li> </ul>
Generic transition team within a paediatric hospital	<p>Involves one or two dedicated nurse specialists who ensure that all young people have appropriate transition support</p>	<p>No current examples</p>
Generic transition services for larger geographical areas	<p>May be more appropriate to relatively rare diseases in Ireland to focus on appropriate co-ordination of care between paediatric services to relevant specialist centres</p>	<ul style="list-style-type: none"> <li>Epidermolysis Bullosa</li> <li>Rare Epilepsy</li> <li>Rare Endocrine disorders</li> <li>Rare Gastroenterology</li> <li>Pituitary disorders and genetic endocrine tumour syndromes Cork University Hospital</li> </ul>

<sup>1</sup> Please note the services listed are examples and this list is not exhaustive.

## 6 RECOMMENDATIONS AND GUIDING PRINCIPLES

### 6.1 INTRODUCTION

Ensuring a safe and effective transition of young people with chronic rare diseases from paediatric care to adult health services is a key quality issue remaining to be addressed nationally.

The UK Child Health Outcomes Forum has recommended two new indicators, one for transition and one for developmentally appropriate healthcare<sup>(91)</sup>. The National Clinical Programme for Rare Diseases considers that this approach should be the cornerstone of transition of care in rare diseases.

The 2012 HIQA National Standards for Safer Better Healthcare Standard 2.3 illustrates the core requirements that are essential to safe transition and integrated care that should be coordinated effectively within and between services, in particular in relation to transition planning. In this, active cooperation between service providers is required, with appropriate sharing of the necessary information to facilitate the safe transfer or sharing of care in a timely and appropriate manner and in line with relevant data protection legislation<sup>(92)</sup>.

The performance of this critical handover of care from the paediatric to the adult setting, including the passing on of all relevant information about the patient and the patient's care, is a core professional responsibility (Recommendation 23.2 of the 2016 Irish Medical Council Guide to Professional Conduct and Ethics for Registered Medical Practitioners)<sup>(93)</sup>.

### 6.2 GUIDING PRINCIPLES FOR TRANSITION IN RARE DISEASES

The National Clinical Programme for Rare Diseases recommends the following guiding principles to ensure a smooth, safe and effective transition of young people with a rare disease from paediatric to adult health services:

1. It is recognised that admission of adolescents/adults to acute adult hospitals may frequently be required to provide urgent/intermittent appropriate medical care to individuals prior to initiation or completion of transition/transfer. However, it should be recognised that this does not constitute the appropriate transition/transfer pathway as outlined in this document to include agreements of acceptance from the appropriate multidisciplinary team and satisfactory medical record transfer. Under these circumstances continuing care post-acute stabilisation should be provided by the ongoing HCP until the transition/transfer process has been completed and agreed.
2. Care should be provided by staff if at all possible with expertise in the management of the particular condition. In line with the recent establishment of European Reference Networks for Rare Disease, adolescents and young adults with rare diseases should receive care if possible at, or in conjunction with, a nationally recognised rare disease health care provider (i.e. a centre of expertise). All cases of rare diseases should be registered with a national centre of expertise, if possible. If there is no national centre of expertise for the condition, the health provider should liaise with a European centre of expertise. Assistance in identifying European expertise can be sought from the National Rare Diseases Office, as Ireland's 'hub' for European Reference Networks. A shared care model between a tertiary centre of expertise and primary care is recommended.

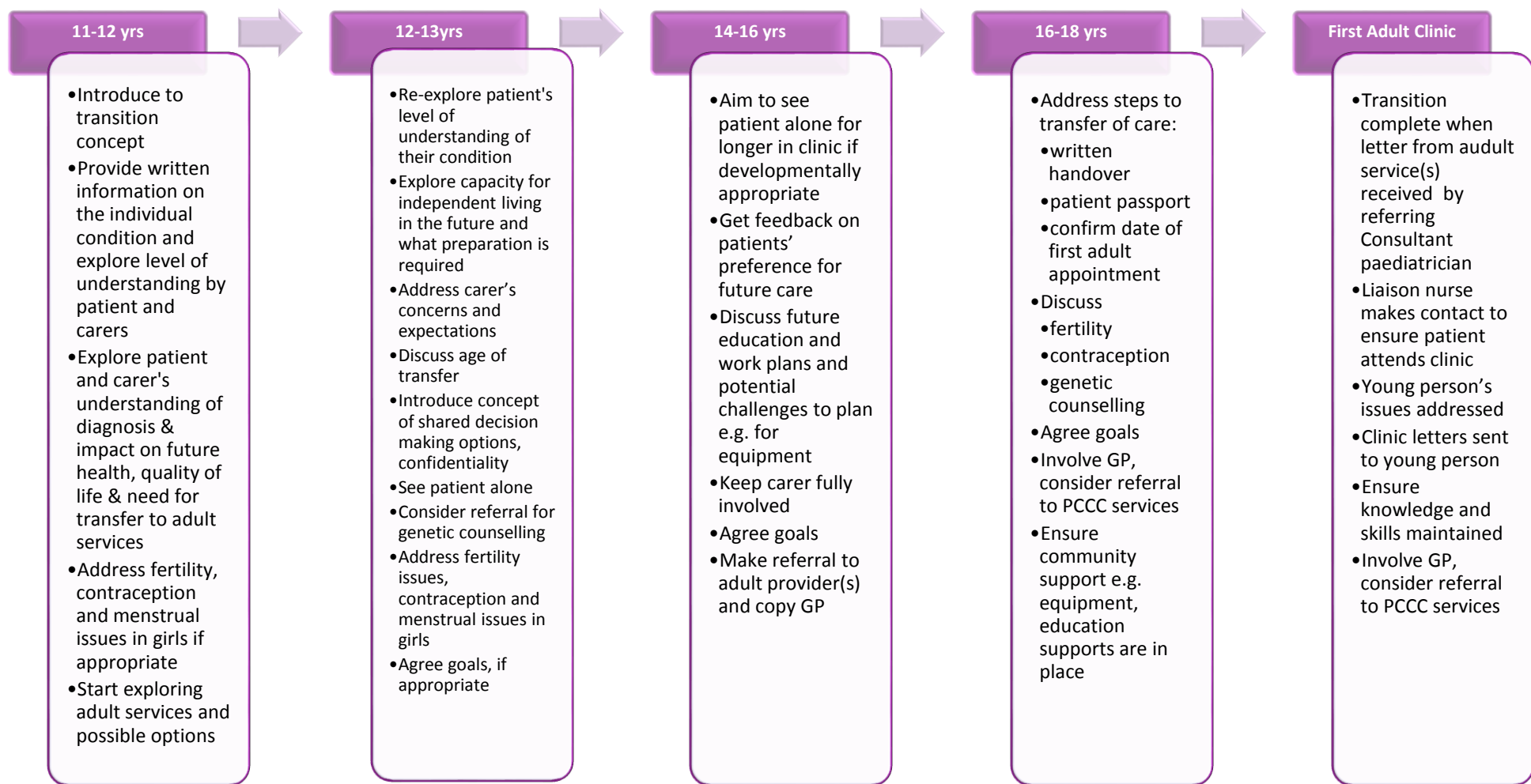
3. Hospitals that provide children/young adults with specialised services for rare diseases should have a policy in place for transition to adult services. This policy should be publically available and should be developed in collaboration with the patient stakeholders. It is recommended that a service specific transition booklet is developed.
4. Paediatric Providers should identify the appropriate adult centre (or multiple centres) of expertise to accept and plan transition planning and build relationships with adult providers to include educational sessions on adolescent concepts.
5. Once the adult provider is agreed, the adult provider should account for the estimated case number to transfer each year and associated costs in the yearly Business Plan.
6. A dedicated transition coordinator should be appointed as a point of contact for each centre. This person should be an experienced health professional (with experience in the particular therapy area or services available) who will have the responsibility for co-ordinating/facilitating transition of care in liaison with the individual clinical teams within the speciality, region or hospital group depending on the condition.
7. Both paediatric and adult providers ideally should share common care pathways and guidelines across sites (to be accessible on the provider website).
8. Paediatric patients, and their families, should be informed of the process of transition well before they start to move their care to adult services. Patients should be given written as well as verbal information about the process in developmentally appropriate language format, and allowing for those with sensory impairments (e.g. Braille or audio). Young adults should be encouraged to participate in evaluating their self-management and setting their own treatment goals, ensuring that the goals are realistic and focused on providing safe, high quality care.
9. The ideal age for patients to transition will depend on the particular condition and co-morbidities and needs to consider the psychological maturity as well as chronological age. Transition should ideally occur between ages 16 to 18 but exceptions may be required for highly complex cases and there is no defined age of transfer in the section on transition of care. The time/age of transfer should be agreed with the patient/family and carer with agreement with the adult provider.
10. Ensure that adequate genetic counselling has been provided to the individual and family. The care pathway and medical summary should be available for each patient (and be provided to the patient), to include a lay person summary of the condition and the most recent updated emergency care plan in developmentally appropriate language format, and allowing for those with sensory impairments (e.g. Braille or audio).
11. Ensure the appropriate education regarding reproductive health and menstrual issues have been addressed commencing from age 12 – 13 or earlier if indicated (See Flowsheet 1).
12. Transition clinics ideally should be held in a paediatric rather than adult setting (if feasible). If not possible, then in an adult setting in an appropriate environment that takes account of the patient's developmental stage and needs, or in a shared clinic with the named adult provider before transition. The carer or family should attend at these clinics. There should be a minimum of one transition clinic visit.
13. At the transition clinic, doctors, nurses, dieticians, clinical psychologists (if possible/ appropriate (and Medical Social Worker, if relevant) and additional MDT team members from both the paediatric and adult teams should attend and ensure comprehensive transfer of patient information to the adult team. Where possible an electronic version should be available for the

patient to have available when/wherever they present acutely/unexpectedly to ensure that key information is presentable to treating physicians.

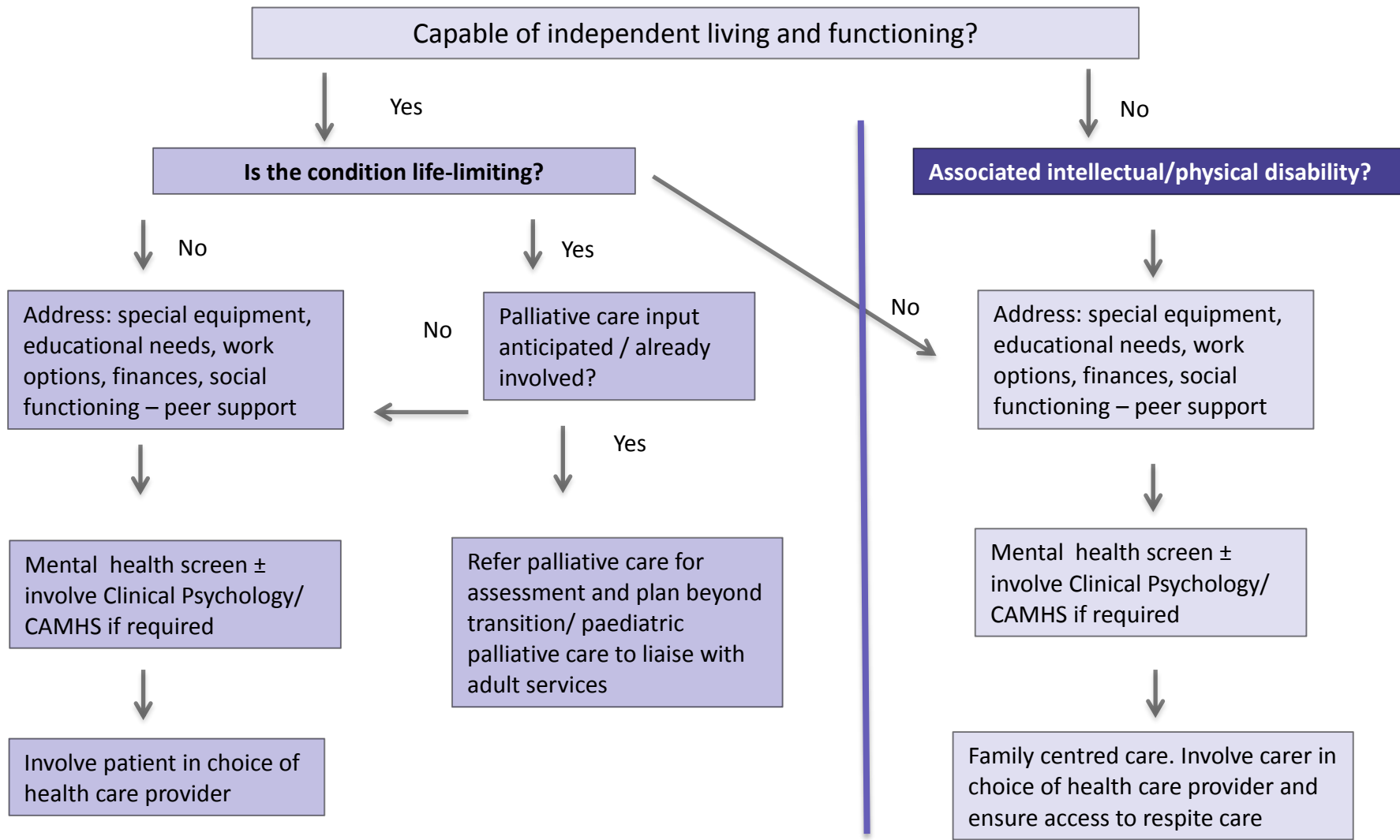
14. The date of first adult OPD appointment is confirmed. The transfer package including medical/surgical, nursing and HSCP summary and care plans (as above), should be transferred to adult provider at least four weeks in advance of the adult OPD appointment.
15. If the patient has a multisystem condition, agreement should be reached as to what is the main system (e.g. neurological, endocrine, and rheumatologic) affected and the appropriate physician to manage that system should be the co-ordinator of the ongoing care into adulthood.
16. A transition Flow Sheet can be used as a guide to support patients and their families to prepare for transition-see appendix 1 for flow sheets.
17. Specific developmentally appropriate discussion and management of contraception/ fertility options (including management of puberty/menstrual issues in adolescents with neurodevelopmental disability) depending on the clinical condition should be offered to all adolescents prior to and during the transition period.
18. Physicians, nurses, health and social care professionals (HSCPs) offering outreach services from centres of expertise to affiliated centres as visiting consultants should be supported so that patients can be offered the best possible care and expertise as close to home as possible. This includes resourcing of multi-disciplinary aspects of the visiting consultant service such as clinical nurse specialist and administrative support.
19. Regular audits of the transition process should be carried out, as well as review of patient feedback, ensuring that patients are not lost to follow up and that they are satisfied with the transition experience. Clinic attendance rates should be evaluated on a regular basis.



**FLWSHEET 1: GUIDING PRINCIPLES FOR TRANSITION OF YOUNG PEOPLE WITH A RARE DISEASE ACCORDING TO AGE**



**FLWSHEET 2: ISSUES TO CONSIDER IN RARE DISEASE TRANSITION FOR A CHILD WITH COMPLEX NEEDS**



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